RGS9 gene

regulator of G-protein signaling 9

Normal Function

The *RGS9* gene provides instructions for making two versions (isoforms) of the RGS9 protein, known as RGS9-1 and RGS9-2. They are found in different parts of the nervous system and have very different functions.

RGS9-1 is produced in the retina, which is the specialized tissue at the back of the eye that detects light and color. Within the retina, RGS9-1 is associated with light-detecting cells called photoreceptors. When light enters the eye, it stimulates specialized pigments in these cells. This stimulation triggers a series of chemical reactions that produce an electrical signal, which is interpreted by the brain as vision. (This process is known as phototransduction.) Once photoreceptors have been stimulated by light, they must return to their resting state before they can be stimulated again. RGS9-1 is involved in a chemical reaction that helps return photoreceptors to their resting state quickly after light exposure.

RGS9-2 is found primarily in an area deep within the brain called the striatum. Although its exact role is unknown, RGS9-2 appears to be part of signaling pathways involving a chemical messenger (neurotransmitter) called dopamine. These pathways are important for planning and coordinating movement. Studies suggest that RGS9-2 also plays a role in the brain's response to opioid drugs, such as morphine and cocaine.

Health Conditions Related to Genetic Changes

bradyopsia

At least two mutations in the *RGS9* gene have been found to cause bradyopsia, a rare condition that affects vision. In people with bradyopsia, the eyes adapt more slowly than usual to changing light conditions (for example, walking out of a darkened movie theater into daylight or driving into a dark tunnel on a sunny day). Some affected individuals also have difficulty seeing small moving objects, such as a tennis ball.

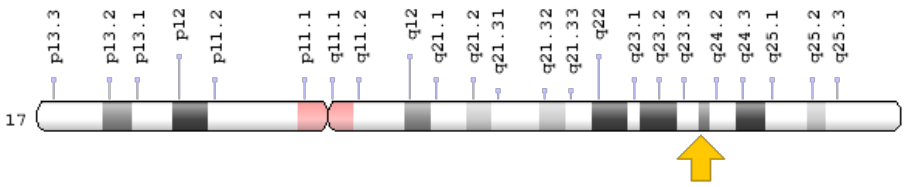
The *RGS9* gene mutations that cause bradyopsia greatly reduce or eliminate the function of RGS9-1 in photoreceptors. (These mutations do not appear to affect the function of RGS9-2 in the brain.) A loss of RGS9-1 function prevents photoreceptors from recovering quickly after responding to light. Normally they return to their resting state in a fraction of a second, but in people with *RGS9* gene mutations, it can take ten seconds or longer. During that time, the photoreceptors cannot respond to light.

This delay causes temporary blindness in response to changing light conditions and may interfere with seeing small objects when they are in motion.

Chromosomal Location

Cytogenetic Location: 17q24.1, which is the long (q) arm of chromosome 17 at position 24.1

Molecular Location: base pairs 65,137,338 to 65,227,703 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MGC111763
- MGC26458
- regulator of G-protein signalling 9
- RGS9L

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: RGS9-1 Phosphorylation and Ca2+ https://www.ncbi.nlm.nih.gov/books/NBK6414/
- Webvision: The Organization of the Retina and Visual System (2010): Phototransduction in Rods and Cones https://www.ncbi.nlm.nih.gov/books/NBK52768/
- Webvision: The Organization of the Retina and Visual System (updated 2012): Photoreceptors https://www.ncbi.nlm.nih.gov/books/NBK11522/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RGS9%5BTIAB%5D%29+OR+%28regulator+of+G-protein+signaling+9%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- REGULATOR OF G PROTEIN SIGNALING 9
  http://omim.org/entry/604067

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9%5Bgene%5D
- HGNC Gene Family: Regulators of G-protein signaling
  http://www.genenames.org/cgi-bin/genefamilies/set/720
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10004
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8787
- UniProt
  http://www.uniprot.org/uniprot/O75916

Sources for This Summary

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